

Health Care Provider Fact Sheet

Disease Name**Cystic Fibrosis****Acronym**

CF

Disease Classification

Genetic Disorder

Formerly known as cystic fibrosis of the pancreas, this entity has increasingly been labeled simply 'cystic fibrosis.' Manifestations relate not only to the disruption of exocrine function of the pancreas but also to intestinal glands (meconium ileus), biliary tree (biliary cirrhosis), bronchial glands (chronic bronchopulmonary infection with emphysema), and sweat glands (high sweat electrolyte with depletion in a hot environment). Infertility occurs in males and females.

Symptom onset

Usually within the first year of life. A small number, however, are not diagnosed until age 18 or older. These patients usually have a milder form of the disease.

Symptoms

Infants with CF have a variety of symptoms including: meconium ileus, liver disease, pancreatic insufficiency, pulmonary disease or an excessive appetite but poor weight gain; and greasy, bulky stools. Symptoms vary from person to person due, in part, to the more than 1,000 mutations of the CF gene. The sweat test is the standard diagnostic test for CF. A sweat test should be performed at a CF Foundation-accredited care center where strict guidelines are followed to ensure accurate results. This simple and painless procedure measures the amount of salt in the sweat. A high salt level indicates CF.

Treatment

The treatment of CF depends upon the stage of the disease and the organs involved. Clearing mucus from the lungs is an important part of the daily CF treatment regimen. In addition, approximately 90 percent of all people with CF take pancreatic enzyme supplements to help them absorb food in digestion.

Emergency Medical Treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website:
<http://www.acmg.net/StaticContent/ACT/CF.pdf>

Inheritance

Autosomal recessive

General population incidence

1 in 2,500 white live births and 1 in 17,000 African American live births.

OMIM Link<http://www3.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=219700>**Genetests Link**www.genetests.org**Support Group**

Cystic Fibrosis Foundation
<http://www.cff.org/home>

National Organization for Rare Diseases
<http://www.rarediseases.org>

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